PARENT FACT SHEET

DISORDER

Carnitine Acylcarnitine Translocase Deficiency (CACT)

CAUSE

CAT deficiency occurs when an enzyme called "carnitine acylcarnitine translocase" is either missing or not working properly. This enzyme's job is to help change certain fats in the food we eat into energy. It also helps to break down fat already stored in the body.

IF NOT TREATED

The effects of this condition vary from person to person. Symptoms can start in infancy, or in milder cases, not until later in childhood. CAT deficiency can cause episodes of illness called metabolic crisis. Some of the first symptoms of metabolic crisis are: extreme sleepiness, behavior changes, irritable mood and poor appetite. Newborns with CAT deficiency often show symptoms within the first week of life. High levels of ammonia in the blood can occur. This can cause serious brain damage. Newborns with CAT may also have: low muscle tone, enlarged liver, heart problems and breathing problems.

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is needed throughout life.

- Infants and young children with CAT need to eat frequently (generally every 4-6 hours) to prevent hypoglycemia or a metabolic crisis.
- A low-fat, low-protein, high-carbohydrate diet is often advised. A dietician will help you create a food plan that meets your child's needs.
- Some children and adults with CAT are helped by taking daily L-carnitine or MCT oil supplements. Your metabolic specialist will prescribe these medications, if necessary.
- Contact your child's doctor immediately at the start of any illness.

IF TREATED

Prompt and careful treatment may help prevent or control symptoms in children with CAT deficiency. However some children continue to have metabolic crisis and other health problems despite treatment. CAT can be a life threatening condition, especially in newborns with symptoms.